

SEVERE CHRONIC

NEUTROPENIA

International Registry

**THE 31<sup>st</sup> LOCAL LIAISON PHYSICIAN MEETING  
OF THE EUROPEAN BRANCH OF THE  
SEVERE CHRONIC NEUTROPENIA INTERNATIONAL REGISTRY**

JOINT WITH

**THE EARLY CAREER INVESTIGATORS MEETING (ECI)  
OF THE COST ACTION EUNET-INNOCHRON**

**14<sup>th</sup> – 16<sup>th</sup> of September 2023  
Verona, VR, Italy**

**Meeting Agenda**



**Venue:**

**Azienda Ospedaliera Universitaria Integrata di Verona, Verona, Italy**

# The 31<sup>st</sup> Local Liaison Physician Meeting

September 14<sup>th</sup> – 16<sup>th</sup> 2023

Verona, VR, Italy

## Friday, September 15<sup>th</sup> 2023

**8:30** (15min) **Welcome and Introduction to the LLP Meeting**

*Cornelia Zeidler and XXXXi*

**8:45** (45min) **Update from the European SCNIR**

*Cornelia Zeidler*

**9:30** (30min) **ELANE genotype-phenotype (TALK1)**

*Cornelia Zeidler*

**10:00** (30min) **ELANE-CyN (TALK2)**

*Karl Welte and Alexander Zeidler*

**10:30** (30min) **SDS**

*Sabine Mellor-Heineke*

## 11:00 Coffee Break

**11:30** (30min) **Implications of newly identified gene mutations in CN**

**12:00** (30min) **Severe phenotype of digenic mutations**

*Karl Welte*

**12:30** (30min) **HAX-1 associated gonadal insufficiencies**

*Denys Pogozhykh*

**13:00** (30min) **New insights in the pathomechanisms of COPZ1 mutation induced CN**

*Natalia Alejandra Borbarán Bravo*

## 13:30 Lunch Break

## 14:30 Eunet-INNOCHRON ECI meeting

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## Saturday, September 16<sup>th</sup> 2023

**09:00** (30min) **Gene corrections**

*Julia Skokowa*

**09:30** (30min) **Drug repurposing in CN**

*Benjamin Dannenmann*

**10:00** (30min) **Drug repurposing in SDS**

*Valentino Bezzerra/Alan Warren*

### 10:30 Coffee Break

**11:00** (30min) **What do we learn from somatic mutations in CN?**

*Julia Skokowa*

**11:30** (60min) **Discussion: 1) Implication of somatic mutations on CN management**

*ALL*

**12:30** (60min) **Discussion 2) New treatment modalities: Indication for gene correction in congenital neutropenia**

*ALL*

### 13:30 Lunch Break

**14:30** (120min) **Case reports-LLP Country Update**

*ALL*

### 16:30 Coffee Break

**17:00** **SCNIR Business**

*ALL*

- How to assess the risk of leukemic transformation in SCN patients?
- Biobank
- Cost Action
- SCNIR Projects

# **The 31<sup>st</sup> Local Liaison Physician Meeting**

September 14<sup>th</sup>– 16<sup>th</sup> 2023

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- Data protection
- SDS Registry
- ASH Abstracts
- LLP Meeting 2024

**17:45**

## **Closing Remarks**

*Cornelia Zeidler, Helen Papadaki*

**18:00 Adjournment**

**Local Organizer: Valentino Bezzeri**